

Supplemental Information

SUPPLEMENTAL TABLE 4 Case Definitions for DBMD for MD STARnet

Case Definition	
Definite	Documented clinical symptoms referable to a dystrophinopathy and direct support of the diagnosis by at least 1 of the following criteria: (1) DNA analysis demonstrating a dystrophin mutation. (2) A muscle biopsy demonstrating abnormal dystrophin, by either immunostaining or Western blot analysis. If there is a family history demonstrating X-linked inheritance, no additional information is required on the muscle. If the diagnosis is based solely on Western blot analysis of muscle biopsy and clinical symptoms, then there must be a description that allows confirmation that adequate numbers of intact muscle fibers were present in the sample to interpret the dystrophin results. If the diagnosis is based on immunostaining, supporting evidence is required from additional stains, for example, utrophin, spectrin, neuronal nitric oxide synthase, or dystroglycans. (3) An elevated creatine kinase, an X-linked pedigree, and an affected family member who meets criterion 1 or 2 above.
Probable	Elevated creatine kinase, a family history consistent with an X-linked muscular dystrophy and documented clinical symptoms referable to a dystrophinopathy. Cases that have abnormal dystrophin results on muscle biopsy but lack the data required in the definition of definite cases, above, are called probable.
Possible	Elevated creatine kinase and documented clinical symptoms referable to a dystrophinopathy, but no muscle biopsy data, dystrophin mutation analysis, or family history to support the diagnosis. Cases that do not have creatine kinase information available in the clinical record, and who do not meet criteria for definite case, are called possible.
Asymptomatic	No clinical symptoms referable to a dystrophinopathy, but laboratory results and/or a positive family history that will likely result in the development of a dystrophinopathy phenotype.
Female	Onset of clinical symptoms referable to a dystrophinopathy appearing before age 21 y and either (1) a dystrophin mutation or (2) a muscle biopsy demonstrating abnormal dystrophin as required for definite case.

SUPPLEMENTAL TABLE 5 Prevalence for DMD and BMD (Definite, Probable, Possible, and Asymptomatic Cases; $n = 704$) per 10 000 Boys, Ages 5 to 9 Years, Identified by MD STARnet, 1991–2010

	Quinquennium	Cases	Population	Prevalence	95% CI
All cases	1991–1995	167	778 716	2.14	(1.83–2.50)
	1996–2000	200	869 321	2.30	(1.99–2.64)
	2001–2005	190	879 300	2.16	(1.86–2.49)
	2006–2010	147	942 791	1.56	(1.32–1.83)
Race/ethnicity					
Non-Hispanic white	1991–1995	111	538 981	2.06	(1.69–2.48)
	1996–2000	121	537 239	2.25	(1.87–2.69)
	2001–2005	109	512 206	2.13	(1.75–2.57)
	2006–2010	89	504 201	1.77	(1.42–2.17)
Non-Hispanic black	1991–1995	10	121 965	0.82	(0.39–1.51)
	1996–2000	19	139 199	1.36	(0.82–2.13)
	2001–2005	14	135 329	1.03	(0.57–1.74)
	2006–2010	11	144 476	0.76	(0.38–1.36)
Hispanic	1991–1995	29	88 570	3.27	(2.19–4.70)
	1996–2000	43	141 042	3.05	(2.21–4.11)
	2001–2005	43	176 768	2.43	(1.76–3.28)
	2006–2010	28	220 040	1.27	(0.85–1.84)
Phenotype					
DMD	1991–1995	125	778 716	1.61	(1.34–1.91)
	1996–2000	141	869 321	1.62	(1.37–1.91)
	2001–2005	149	879 300	1.69	(1.43–1.99)
	2006–2010	113	942 791	1.20	(0.99–1.44)
BMD	1991–1995	42	778 716	0.54	(0.39–0.73)
	1996–2000	58	869 321	0.67	(0.51–0.86)
	2001–2005	39	879 300	0.44	(0.32–0.61)
	2006–2010	32	942 791	0.34	(0.23–0.48)

CI, confidence interval.

SUPPLEMENTAL TABLE 6 Prevalence in 2010 for DMD and BMD (Definite, Probable, Possible, and Asymptomatic Cases; $n = 561$) per 10 000 Male Individuals, Ages 5 to 24 Years, Identified by MD STARnet, 1991–2010

	Age Group, y	Cases	Population	Prevalence	95% CI
All cases					
	All	561	3 827 532	1.47	(1.35–1.59)
	20–24	102	960 866	1.06	(0.87–1.29)
	15–19	168	985 263	1.71	(1.46–1.98)
	10–14	161	938 612	1.72	(1.46–2.00)
	5–9	130	942 791	1.38	(1.15–1.64)
Race/ethnicity					
Non-Hispanic white	All	328	2 147 039	1.53	(1.37–1.70)
	20–24	65	564 779	1.15	(0.89–1.47)
	15–19	98	558 178	1.76	(1.43–2.14)
	10–14	88	519 881	1.69	(1.36–2.09)
	5–9	77	504 201	1.53	(1.21–1.91)
Non-Hispanic black	All	44	605 232	0.73	(0.53–0.98)
	20–24	4	143 512	0.28	(0.08–0.71)
	15–19	18	166 175	1.08	(0.64–1.71)
	10–14	11	151 069	0.73	(0.36–1.30)
	5–9	11	144 476	0.76	(0.38–1.36)
Hispanic	All	125	797 913	1.57	(1.30–1.87)
	20–24	22	185 830	1.18	(0.74–1.79)
	15–19	38	192 816	1.97	(1.39–2.71)
	10–14	38	199 227	1.91	(1.35–2.62)
	5–9	27	220 040	1.23	(0.81–1.79)
Phenotype					
DMD	All	410	3 827 532	1.07	(0.97–1.18)
	20–24	69	960 866	0.72	(0.56–0.91)
	15–19	117	985 263	1.19	(0.98–1.42)
	10–14	125	938 612	1.33	(1.11–1.59)
	5–9	99	942 791	1.05	(0.85–1.28)
BMD	All	147	3 827 532	0.38	(0.32–0.45)
	20–24	33	960 866	0.34	(0.24–0.48)
	15–19	50	985 263	0.51	(0.38–0.67)
	10–14	34	938 612	0.36	(0.25–0.51)
	5–9	30	942 791	0.32	(0.21–0.45)

CI, confidence interval.